



## Sequence Revision History

PubMed

Nucleotide

Protein

Genome

Structure

PMC

Taxonomy

OMIM

Find (Accessions, GI numbers or Fasta style SeqIds) **NM\_000485**

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Gi	Version	Update Date
71773149	2	Jun 22 2008 12:01 PM
4502170	1	Sep 6 2003 6:28 PM

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Clusters of orthologous groups

Protein reviews on the web

LOCUS NM\_000485 807 bp mRNA linear PR1  
 DEFINITION Homo sapiens adenine phosphoribosyltransferase (APRT), variant 1, mRNA.  
 LOCUS NM\_000485 841 bp mRNA linear PR1  
 DEFINITION Homo sapiens adenine phosphoribosyltransferase (APRT),  
 ACCESSION NM\_000485  
 VERSION NM\_000485.2 GI:71773149  
 VERSION NM\_000485.1 GI:4502170  
 KEYWORDS .  
 SOURCE Homo sapiens (human)  
 ORGANISM Homo sapiens  
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Eut  
 Mammalia; Eutheria; Euarchontoglires; Primates; Haplor  
 Catarrhini; Hominidae; Homo.  
 REFERENCE 1 (bases 1 to 807)  
 AUTHORS Silva,C.H., Silva,M., Iulek,J. and Thiemann,O.H.  
 TITLE Structural complexes of human adenine phosphoribosyltra  
 reveal novel features of the APRT catalytic mechanism  
 JOURNAL J. Biomol. Struct. Dyn. 25 (6), 589-597 (2008)  
 PUBMED 18399692  
 REMARK GeneRIF: Data indicates that the flexible loop structu  
 open conformation before and after binding of both subs  
 adenine and phosphoribosyl pyrophosphate.  
 REFERENCE 2 (bases 1 to 807)  
 AUTHORS Di Pietro,V., Perruzza,I., Amorini,A.M., Balducci,A.,  
 Ceccarelli,L., Lazzarino,G., Barsotti,P., Giardina,B. a  
 Tavazzi,B.  
 TITLE Clinical, biochemical and molecular diagnosis of a comp  
 homozygote for the 254 bp deletion-8 bp insertion of th  
 suffering from severe renal failure  
 JOURNAL Clin. Biochem. 40 (1-2), 73-80 (2007)  
 PUBMED 17126311  
 REMARK GeneRIF: APRT assay in a sample of patient hemolysate s  
 detectable activity of the enzyme (25.56+/-9.55 U/L rec  
 in control healthy subjects).  
 REFERENCE 3 (bases 1 to 807)  
 AUTHORS Ewing,R.M., Chu,P., Elisma,F., Li,H., Taylor,P., Climie  
 McBroom-Cerajewski,L., Robinson,M.D., O'Connor,L., Li,M  
 Taylor,R., Dharsee,M., Ho,Y., Heilbut,A., Moore,L., Zha  
 Ornatsky,O., Bukhman,Y.V., Ethier,M., Sheng,Y., Vasile  
 Abu-Farha,M., Lambert,J.P., Duewel,H.S., Stewart,I.I.,  
 Hogue,K., Colwill,K., Gladwish,K., Muskat,B., Kinach,R.  
 Adams,S.L., Moran,M.F., Morin,G.B., Topaloglou,T. and I  
 TITLE Large-scale mapping of human protein-protein interactio  
 spectrometry  
 JOURNAL Mol. Syst. Biol. 3, 89 (2007)  
 PUBMED 17353931

EXHIBIT

6a

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REFERENCE 4 (bases 1 to 807)  
AUTHORS Bruneel,A., Labas,V., Mailloux,A., Sharma,S., Royer,N.,  
Pernet,P., Vaubourdolle,M. and Baudin,B.  
TITLE Proteomics of human umbilical vein endothelial cells ap  
etoposide-induced apoptosis  
JOURNAL Proteomics 5 (15), 3876-3884 (2005)  
PUBMED 16130169

REFERENCE 5 (bases 1 to 807)  
AUTHORS Taniguchi,A., Tsuchida,S., Kuno,S., Mita,M., Machida,T.  
Ioritani,N., Terai,C., Yamanaka,H. and Kamatani,N.  
TITLE Identification of two novel mutations in adenine  
phosphoribosyltransferase gene in patients with  
2,8-dihydroxyadenine urolithiasis  
JOURNAL Nucleosides Nucleotides Nucleic Acids 23 (8-9), 1141-11  
PUBMED 15571218  
REMARK GeneRIF: two novel mutations, G133D and V84M, were four  
APRT gene in Japanese patients with APRT deficiency

REFERENCE 6 (bases 1 to 807)  
AUTHORS Kamatani,N., Hakoda,M., Otsuka,S., Yoshikawa,H. and Kas  
TITLE Only three mutations account for almost all defective a  
causing adenine phosphoribosyltransferase deficiency in  
patients  
JOURNAL J. Clin. Invest. 90 (1), 130-135 (1992)  
PUBMED 1353080

REFERENCE 7 (bases 1 to 807)  
AUTHORS Chen,J., Sahota,A., Laxdal,T., Scrine,M., Bowman,S., Cu  
Stambrook,P.J. and Tischfield,J.A.  
TITLE Identification of a single missense mutation in the ade  
phosphoribosyltransferase (APRT) gene from five Iceland  
and a British patient  
JOURNAL Am. J. Hum. Genet. 49 (6), 1306-1311 (1991)  
PUBMED 1746557

REFERENCE 8 (bases 1 to 807)  
AUTHORS Ludwig,H., Kuzmits,R., Pietschmann,H. and Muller,M.M.  
TITLE Enzymes of the purine interconversion system in chronic  
leukemia: decreased purine nucleoside phosphorylase and  
deaminase activity  
JOURNAL Blut 39 (5), 309-315 (1979)  
PUBMED 116697

REFERENCE 9 (bases 1 to 807)  
AUTHORS Holden,J.A., Meredith,G.S. and Kelley,W.N.  
TITLE Human adenine phosphoribosyltransferase. Affinity purifi  
subunit structure, amino acid composition, and peptide  
JOURNAL J. Biol. Chem. 254 (15), 6951-6955 (1979)  
PUBMED 457664

REFERENCE 10 (bases 1 to 807)  
AUTHORS Johnson,L.A., Gordon,R.B. and Emmerson,B.T.  
TITLE Adenine phosphoribosyltransferase: a simple spectrophot  
assay and the incidence of mutation in the normal popul  
JOURNAL Biochem. Genet. 15 (3-4), 265-272 (1977)  
PUBMED 869896

Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Ho  
REFERENCE 1 (bases 1 to 841)  
AUTHORS Wang,L., Ou,X., Sebesta,I., Vondrak,K., Krijt,J., Ellec  
Poupetova,H., Ledvinova,J., Zeman,J., Simmonds,H.A.,  
Tischfield,J.A. and Sahota,A.  
TITLE Combined adenine phosphoribosyltransferase and  
N-acetylgalactosamine-6-sulfate sulfatase deficiency  
JOURNAL Mol. Genet. Metab. 68 (1), 78-85 (1999)  
PUBMED 10479485

**EXHIBIT**

6b

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- REFERENCE 2 (bases 1 to 841)  
 AUTHORS Siegfried,Z., Eden,S., Mendelsohn,M., Feng,X., Tsuberi,  
 Cedar,H.  
 TITLE DNA methylation represses transcription in vivo  
 JOURNAL Nat. Genet. 22 (2), 203-206 (1999)  
 PUBMED 10369268
- REFERENCE 3 (bases 1 to 841)  
 AUTHORS Yagisawa,T., Yamazaki,Y., Toma,H. and Kamatani,N.  
 TITLE Radiopaque 2,8-dihydroxyadenine lithiasis  
 JOURNAL Int Urol Nephrol 31 (2), 141-143 (1999)  
 PUBMED 10481956
- REFERENCE 4 (bases 1 to 841)  
 AUTHORS Menardi,C., Schneider,R., Neuschmid-Kaspar,F., Klocker,  
 Hirsch-Kauffmann,M., Auer,B. and Schweiger,M.  
 TITLE Human APRT deficiency: indication for multiple origins  
 common Caucasian mutation and detection of a novel type  
 involving intrastrand-templated repair  
 JOURNAL Hum. Mutat. 10 (3), 251-255 (1997)  
 PUBMED 9298830
- REFERENCE 5 (bases 1 to 841)  
 AUTHORS Boyadjiev,S.A., Sahota,A. and Tischfield,J.A.  
 TITLE Identification and application of polymorphisms flankin  
 adenine phosphoribosyltransferase gene  
 JOURNAL Hum. Mutat. 8 (3), 214-215 (1996)  
 PUBMED 8889579
- REFERENCE 6 (bases 1 to 841)  
 AUTHORS Tischfield,J.A., Engle,S.J., Gupta,P.K., Bye,S., Boyad  
 Shao,C., O'Neill,P., Albertini,R.J., Stambrook,P.J. and  
 Germline and somatic mutation at the APRT locus of mice  
 TITLE  
 JOURNAL Adv. Exp. Med. Biol. 370, 661-664 (1994)  
 PUBMED 7660991
- REFERENCE 7 (bases 1 to 841)  
 AUTHORS Takeuchi,H., Kaneko,Y., Fujita,J. and Yoshida,O.  
 TITLE A case of a compound heterozygote for adenine  
 phosphoribosyltransferase deficiency (APRT\*J/APRT\*Q0) I  
 2,8-dihydroxyadenine urolithiasis: review of the report  
 with 2,8-dihydroxyadenine stones in Japan  
 JOURNAL J. Urol. 149 (4), 824-826 (1993)  
 PUBMED 8455250
- REFERENCE 8 (bases 1 to 841)  
 AUTHORS Chen,J., Sahota,A., Stambrook,P.J. and Tischfield,J.A.  
 TITLE Polymerase chain reaction amplification and sequence an  
 human mutant adenine phosphoribosyltransferase genes: t  
 and frequency of errors caused by Taq DNA polymerase  
 JOURNAL Mutat. Res. 249 (1), 169-176 (1991)  
 PUBMED 2067530
- REFERENCE 9 (bases 1 to 841)  
 AUTHORS Hidaka,Y., Tarle,S.A., O'Toole,T.E., Kelley,W.N. and Pa  
 TITLE Nucleotide sequence of the human APRT gene  
 JOURNAL Nucleic Acids Res. 15 (21), 9086 (1987)  
 PUBMED 3684585
- REFERENCE 10 (bases 1 to 841)  
 AUTHORS Broderick,T.P., Schaff,D.A., Bertino,A.M., Dush,M.K.,  
 Tischfield,J.A. and Stambrook,P.J.  
 TITLE Comparative anatomy of the human APRT gene and enzyme:  
 sequence divergence and conservation of a nonrandom CpC  
 dinucleotide arrangement  
 JOURNAL Proc. Natl. Acad. Sci. U.S.A. 84 (10), 3349-3353 (1987)  
 PUBMED 3554238
- REFERENCE 11 (bases 1 to 841)

**EXHIBIT**

6c

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AUTHORS Wilson, J.M., O'Toole, T.E., Argos, P., Shewach, D.S., Dadgar, Kelley, W.N.

TITLE Human adenine phosphoribosyltransferase. Complete amino acid sequence of the erythrocyte enzyme

JOURNAL J. Biol. Chem. 261 (29), 13677-13683 (1986)

PUBMED 3531209

COMMENT REVIEWED REFSEQ: This record has been curated by NCBI. The reference sequence was derived from BM423481.1 and BU506. On Aug 3, 2005 this sequence version replaced gi:450217. The reference sequence was derived from Y00486.1.

Summary: Adenine phosphoribosyltransferase belongs to the purine/pyrimidine phosphoribosyltransferase family. A characteristic feature of this gene is the distribution of CpG dinucleotides. The enzyme catalyzes the formation of AMP and inorganic pyrophosphate from adenine and 5-phosphoribosyl-1-pyrophosphate (PRPP). The enzyme produces adenine as a by-product of the polyamine biosynthesis pathway. A homozygous deficiency in this enzyme causes 2,8-dihydroxyadenine urolithiasis. Two transcript variants encoding different isoforms have been found for this gene.

Transcript Variant: This variant (1) represents the longer transcript and encodes the longer isoform (a).

Publication Note: This RefSeq record includes a subset of publications that are available for this gene. Please see the Entrez Gene record to access additional publications.

COMPLETENESS: complete on the 3' end.

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	714-807	BU507629.1	115-208
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EXHIBIT

6d

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## ORIGIN

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841  c

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**EXHIBIT**

6e

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